

**Michigan Public Health Institute  
Region 4 Priority 2 Workgroup Quarterly report  
Inborn Errors of Metabolism - Information System  
June 1 2008 to August 312008**

**Project Director: Cynthia Cameron; Senior Project Coordinator: Sally J. Hiner  
Project Co-Leads: Susan A. Berry, MD & Carolyn Anderson**

**Region 4 Progress Review for Priority 2 Workgroup (Grant began in June, 2007)**

Workgroup communication and membership

- Clinical representatives from Iowa and Nebraska in the Heartland Region were added to the workgroup.
- Workgroup Facilitator worked with Co-Leads to plan, prepare and disseminate materials for and convene monthly telemeetings of the workgroup.

Region 4 Metabolic Center IRB Progress and Training

- Three (3) centers, Wayne State University in Detroit, MI, Nationwide Children's Hospital in Columbus, Ohio and Case Western Reserve in Ohio, obtained IRB approval this quarter.
- Centers with IRB approval pending include Waisman Center in Madison, WI, University of Kentucky Louisville, and University of Iowa in the Heartland Region.
- State clinical leads are collaborating with additional metabolic centers in their respective states to promote the IRB process at centers in their state as an initial step toward their full participation in the IBEM-IS.
- Expertise and assistance is shared among work group members to support the IRB process including templates, narratives, other resources and suggestions. IRB documents available on the Region 4 website for ease of access were updated this quarter.
- One session to train users in entering metabolic patients into the IBEM-IS DocSite system was conducted this quarter by Susan Berry and Kristi Bentler.
- Workgroup facilitator continued tracking IRB approvals, responding to requests for technical assistance with IRB applications, obtaining and processing requests for user names and passwords and coordinated web-based training for eligible sites.

IBEM- IS Disorder Expansion Update

- The workgroup continued activities to add metabolic disorders to the information system. This quarter the group drafted outcome measures (data elements) for one new disorder, Galactosemia and refined the draft of data elements for Glutaric Acidemia Type 1 (GA Type 1). (This brings the total number disorders for which we have sets of data elements for enrollment and interval visits to thirty (30) including: fatty acid oxidation disorders of CACT, CPT-1, CPT-2, LCHAD, MCAD, Primary Carnitine Transporter, SCAD, Trifunctional Protein deficiency (TFP), and VLCAD; Amino acid disorder and Maple Syrup Urine Disease (MSUD); and Organic acid disorders of Beta-ketothiolase, 3-MCC, 3-MGA Type 1, Homocystinuria, HMG CoA Lyase, Isobutyryl CoA dehydrogenase deficiency (IBD), 3-methyl 3-OH butyryl CoA dehydrogenase, succinyl CoA-3-ketoacid transferase deficiency (SCOT), MMA (Mut 0)

MMA (Mut -), MMA (cblA), MMA (cblC), MMA (cblD variant 2), MMA (cbl F), MMA (transcobalamin II), Propionic acidemia. Biotinidase deficiency is also complete.)

- Workgroup facilitator continues to serve as liaison between IBEM-IS workgroup and DocSite to facilitate the addition of enrollment and interval surveys for a new disorder approximately every three to four weeks.
- Sue Berry and Kristi Bentler continue to extensively review the information system surveys as they are added to assure continuing accurate, consistent and comprehensive data collection. This intensive review activity is recorded on “DocSite Communication Worksheets”. The worksheets are forwarded to DocSite and phone and email contact, facilitated and coordinated by the Senior Project Coordinator, continues in order to accomplish the goal of adding metabolic disorders to the platform and entering patients.
- Anne Jurek, Project Epidemiologist, is working directly with DocSite staff to facilitate reporting mechanisms for exporting data.

#### Patient enrollment into IBEM-IS

- As of July 31, 40 patients were enrolled in the information system, 37 with MCAD and 3 with MSUD.
- As of July 31, one hundred forty one (141) patients with a variety of disorders such as CPT-1, CPT-2, MCAD, LCHAD, TFP, VLCAD have signed consents. With the addition of these disorders to the DocSite platform this quarter, these patients are now being added to the IBEM-IS.
- An IBEM-IS case log form was drafted to track and update both the number of individuals consented to participate in the IBEM-IS and those that have been enrolled. Information is recorded by disorder. This log will be useful in obtaining cumulative numbers of both patients consented by the clinical centers and patients entered into the information system as the system moves forward.

#### Region 4 web site enhancement

- Workgroup facilitator requested and posted updates to all materials available on the IBEM-IS workgroup page. The site offers a method of sharing resources, forms, materials, enrollment and interval surveys and updates. A meeting calendar, agendas and summaries are posted as well as metabolic disorder surveys as they are developed. Updated work group contact information can be maintained. The IBEM-IS Work Group is afforded an infrastructure to communicate efficiently and effectively prior to and between meetings.

#### IBEM-IS Collaboration beyond Region 4

- New initiatives and partnerships strengthened by grant submission: Partnership with NIH Consortium to promote a regional and consortium-wide data base for patients with Fatty Acid Oxidation Disorders (FAOD)
  - The FAOD Clinical Research and Treatment Network (FAODCRTN) partnered with Region 4 IBEM-IS Priority 2 Project to adopt the IBEM-IS database in order to input and access data for an increased number of patients with FAOD. Further, this offers a means to perform longitudinal research and to identify patients for

- clinical studies. Functionally, the Priority 2 workgroup will maintain as much of the established infrastructure as possible while expanding to allow others to participate. This consortium would be a part of the IBEM-IS Priority 2 Work Group.
- Efforts to extend IBEM-IS participation beyond the region is now in process. Nebraska and Iowa are participating in the Region 4 Priority 2 IBEM-IS Workgroup telemeetings. Both state sites are in the process of completing IRB applications for participation in case entry.
  - Susan Berry has agreed to chair the medical foods expert workgroup for the Secretary's Advisory Committee. Region 4 will participate in a national survey to ask parents about coverage for medical foods, nutraceuticals, and medical equipment. Dr. Berry attended a meeting of this group in June 2008 to begin development of plans for improvement of coverage for nutritional treatments for children affected with inborn errors of metabolism. Dr. Berry and Dr. Cameron, Project Director, continue to work with this group via telemeetings and email communication.
  - Dr. Berry was invited to participate in the NYMAC project to develop standardization for diagnostic following ascertainment with newborn blood spot screening. She attended this workgroup in June 2008.

### Expanding IBEM-IS

#### Endocrine Workgroup

- IBEM-IS and Region 4 staff continue support for efforts of the endocrine work group to expand the IBEM-IS to include data elements for congenital adrenal hyperplasia, a rare serious disorder found by newborn screening.

### **Difficulties encountered and solutions**

- Participation of short and long term follow up staff from the state departments of health has been inconsistent. Plans are underway to obtain input from the state staff about their role with the workgroup, how they can contribute to the group and what is important for them to gain. This will include exploring their data needs related to short term and long term outcomes via an informal survey.
- Communication methods with DocSite staff have been revised and formalized and have improved; we will continue to evaluate the effectiveness of this communication as we go forward.

### **Changes in key personnel**

None

### **Planned Activities**

- IBEM-IS work group will meet face to face in September, 2008 in Lansing, MI to move work of the project forward.
- Research proposals and ideas will be discussed and encouraged. A review process for specific proposals is in place.

- Work group will refine neurologic data elements for GA1 and develop surveys (data elements) for galactosemia. New disorder elements will be added to the IBEM-IS via work with DocSite.
- Workgroup plans ongoing evaluation of recent documents and accomplishments at monthly phone conference calls.
- Active patient enrollment in the IBEM-IS will continue and increase.
- Training for data entry will be accomplished with new participating centers.
- Monitor progress of implementing MEMSCIS as a clinical tool within the workgroup
- Continue collaborations with FAODCRTN and with key staff from Iowa and Nebraska who are now participating from the Heartland Region in IBEM-IS project.